METHODS OF INHERITANCE

Genetic Disorders and How They are Passed from Generation to Generation

Chromosomal Disorders

Genetic disorders can occur when there is too much or too little chromosomal material or when an abnormal rearrangement (translocation) is present. Chromosomes, which are structural elements containing genes, normally occur in 23 pairs. The first 22 pairs are called autosomes and the last pair is called the sex pair — XX (female) or XY (male).

Examples: Down Syndrome, Turner Syndrome.

Single Gene Disorders

Genetic disorders also can result when an alteration occurs in a gene. Genes carry the bits of information which determine the development and function of an individual. Half of our genes are inherited from our mother and half from our father.

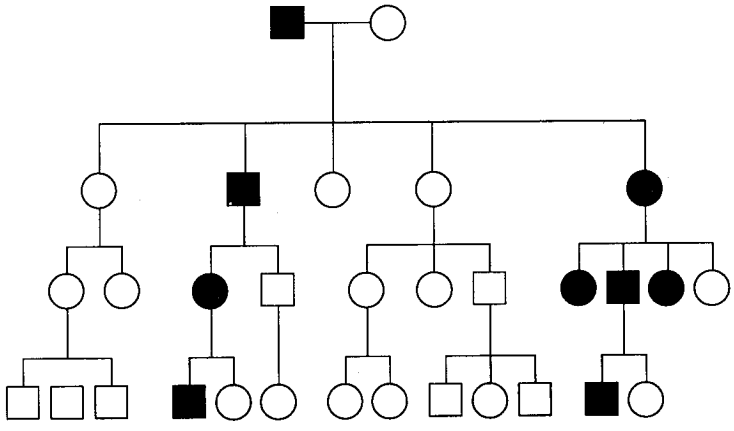
Patterns of Single Gene Disorder Inheritance

Autosomal Dominant - ("vertical inheritance") - a gene from one parent is faulty and its action dominates the other gene in the pair (inherited from the other parent) and may cause a disorder.

- The disorder shows up in the presence of one abnormal gene.
- Several generations may be affected or the disorder may occur as a mutation.
- For an affected person, there is a 50% risk in each pregnancy that offspring will be affected.
- Occurs with equal frequency in both sexes.

Examples: Marfan Syndrome, Achondroplasia, Ectrodactyly.

A typical autosomal dominant pedigree:

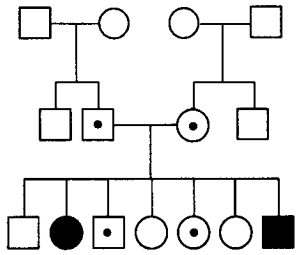


Autosomal Recessive - ("horizontal inheritance") - the genes from both parents must be faulty to cause the individual to have the disorder.

- Both parents carry one abnormal gene, but are physically and mentally normal.
- Gene carriers who are unaffected may exist in many generations.
- If both parents are carriers, there is a 25% risk in each pregnancy that offspring will be affected with the disorder.
- Occurs with equal frequency in both sexes.

Examples: Albinism, Sickle Cell, Tay Sachs, Cystic Fibrosis, Diastrophic Dwarfism, PKU.

A typical autosomal recessive pedigree:



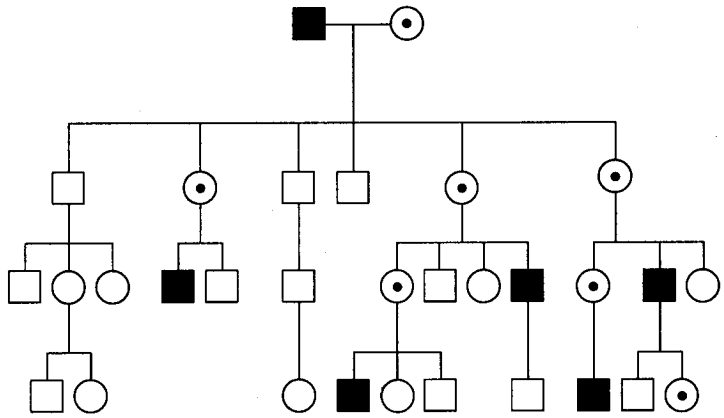
X-linked - A faulty gene is located on the X chromosome.

If female is carrier: 50% risk for sons being affected;
50% risk for daughters being carriers.

If male has abnormal gene: 100% risk for daughters being carriers;
Sons not affected as they receive only the Y chromosome.

Examples: Color Blindness, Fragile X, Ectodermal Dysplasia, Hemophilia.

A typical X-linked recessive pedigree:



MULTIFACTORIAL

A multifactorial disorder results from an interaction of genes from both parents and environmental influences.

Examples: Cleft Lip and/or Palate, Spina Bifida

TERATOGENIC

Teratogenic disorders result from environmental agents which may adversely affect fetal development.

Example: Drugs and alcohol, maternal infections.

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